



ring chromosome 14 syndrome

Ring chromosome 14 syndrome is a condition characterized by seizures and intellectual disability. Recurrent seizures (epilepsy) develop in infancy or early childhood. In many cases, the seizures are resistant to treatment with anti-epileptic drugs. Most people with ring chromosome 14 syndrome also have some degree of intellectual disability or learning problems. Development may be delayed, particularly the development of speech and of motor skills such as sitting, standing, and walking.

Additional features of ring chromosome 14 syndrome can include slow growth and short stature, a small head (microcephaly), puffy hands and/or feet caused by a buildup of fluid (lymphedema), and subtle differences in facial features. Some affected individuals have problems with their immune system that lead to recurrent infections, especially involving the respiratory system. Abnormalities of the retina, the specialized tissue at the back of the eye that detects light and color, have also been reported in some people with this condition. These changes typically do not affect vision. Major birth defects are rarely seen with ring chromosome 14 syndrome.

Frequency

Ring chromosome 14 syndrome appears to be a rare condition, although its prevalence is unknown. More than 50 affected individuals have been reported in the medical literature.

Genetic Changes

Ring chromosome 14 syndrome is caused by a chromosomal abnormality known as a ring chromosome 14, sometimes written as r(14). A ring chromosome is a circular structure that occurs when a chromosome breaks in two places and its broken ends fuse together. People with ring chromosome 14 syndrome have one copy of this abnormal chromosome in some or all of their cells.

Researchers believe that several critical genes near the end of the long (q) arm of chromosome 14 are lost when the ring chromosome forms. The loss of these genes is likely responsible for several of the major features of ring chromosome 14 syndrome, including intellectual disability and delayed development. Researchers are still working to determine which missing genes contribute to the signs and symptoms of this disorder.

Epilepsy is a common feature of ring chromosome syndromes, including ring chromosome 14. There may be something about the ring structure itself that causes epilepsy. Seizures may occur because certain genes on the ring chromosome 14 are

less active than those on the normal chromosome 14. Alternately, seizures might result from instability of the ring chromosome in some cells.

Inheritance Pattern

Ring chromosome 14 syndrome is almost never inherited. A ring chromosome typically occurs as a random event during the formation of reproductive cells (eggs or sperm) or in early embryonic development. In some cases, the ring chromosome is present in only some of a person's cells. This situation is known as mosaicism.

Most affected individuals have no history of the disorder in their families. However, at least two families have been reported in which a ring chromosome 14 was passed from a mother to her children.

Other Names for This Condition

- ring 14
- ring 14 syndrome
- ring chromosome 14

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Ring chromosome 14
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2930916/>

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Chromosome
<https://medlineplus.gov/ency/article/002327.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Chromosome
<https://medlineplus.gov/ency/article/002327.htm>
- Health Topic: Developmental Disabilities
<https://medlineplus.gov/developmentaldisabilities.html>
- Health Topic: Epilepsy
<https://medlineplus.gov/epilepsy.html>

Genetic and Rare Diseases Information Center

- Ring chromosome 14
<https://rarediseases.info.nih.gov/diseases/6072/ring-chromosome-14>

Additional NIH Resources

- National Human Genome Research Institute: Chromosome Abnormalities
<https://www.genome.gov/11508982/>
- National Institute of Neurological Disorders and Stroke: Epilepsy Information Page
<https://www.ninds.nih.gov/Disorders/All-Disorders/Epilepsy-Information-Page>

Educational Resources

- MalaCards: ring chromosome 14 syndrome
http://www.malacards.org/card/ring_chromosome_14_syndrome
- March of Dimes: Chromosomal Conditions
<http://www.marchofdimes.org/baby/chromosomal-conditions.aspx>
- Orphanet: Ring chromosome 14
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1440
- Unique support group booklet about ring chromosome 14
<http://www.rarechromo.org/information/Chromosome%2014/Ring%2014%20FTNW.pdf>

Patient Support and Advocacy Resources

- American Epilepsy Society
<https://www.aesnet.org/>
- Associazione Internazionale Ring 14 (Ring 14 International Support Group)
<http://www.ring14.org/eng/>
- Chromosome Disorder Outreach
<http://chromodisorder.org/>

- Citizens United for Research in Epilepsy (CURE)
<http://www.cureepilepsy.org/home.asp>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/chromosome-14-ring/>
- Unique - The Rare Chromosome Disorder Support Group (UK)
<http://www.rarechromo.org/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ring+chromosome+14+syndrome%5BTIAB%5D%29+OR+%28ring+14+syndrome%5BTIAB%5D%29+OR+%28ring+chromosome+14%5BTIAB%5D%29+OR+%28ring+14%5BTIAB%5D%29+NOT+%28benzene%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

Sources for This Summary

- Giovannini S, Marangio L, Fusco C, Scarano A, Frattini D, Della Giustina E, Zollino M, Neri G, Gobbi G. Epilepsy in ring 14 syndrome: a clinical and EEG study of 22 patients. *Epilepsia*. 2013 Dec;54(12):2204-13. doi: 10.1111/epi.12393. Epub 2013 Oct 1.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24116895>
- Morimoto M, Usuku T, Tanaka M, Otabe O, Nishimura A, Ochi M, Takeuchi Y, Yoshioka H, Sugimoto T. Ring chromosome 14 with localization-related epilepsy: three cases. *Epilepsia*. 2003 Sep;44(9):1245-9.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12919399>
- Schlade-Bartusiak K, Costa T, Summers AM, Nowaczyk MJ, Cox DW. FISH-mapping of telomeric 14q32 deletions: search for the cause of seizures. *Am J Med Genet A*. 2005 Oct 15;138A(3):218-24.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16152642>
- Specchio N, Trivisano M, Serino D, Cappelletti S, Carotenuto A, Claps D, Marras CE, Fusco L, Elia M, Vigeveno F. Epilepsy in ring 14 chromosome syndrome. *Epilepsy Behav*. 2012 Dec;25(4):585-92. doi: 10.1016/j.yebeh.2012.09.032. Epub 2012 Nov 14. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23159383>
- Unique support group booklet about ring chromosome 14
<http://www.rarechromo.org/information/Chromosome%2014/Ring%2014%20FTNW.pdf>
- Zollino M, Ponzi E, Gobbi G, Neri G. The ring 14 syndrome. *Eur J Med Genet*. 2012 May;55(5):374-80. doi: 10.1016/j.ejmg.2012.03.009. Epub 2012 Apr 14. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22564756>

- Zollino M, Seminara L, Orteschi D, Gobbi G, Giovannini S, Della Giustina E, Frattini D, Scarano A, Neri G. The ring 14 syndrome: clinical and molecular definition. *Am J Med Genet A*. 2009 Jun; 149A(6):1116-24. doi: 10.1002/ajmg.a.32831.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19441122>
 - van Karnebeek CD, Quik S, Sluijter S, Hulsbeek MM, Hoovers JM, Hennekam RC. Further delineation of the chromosome 14q terminal deletion syndrome. *Am J Med Genet*. 2002 Jun 1; 110(1):65-72. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12116274>
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